



WNT10A gene

Wnt family member 10A

Normal Function

The *WNT10A* gene is part of a large family of WNT genes, which play critical roles in development starting before birth. These genes provide instructions for making proteins that participate in chemical signaling pathways in the body. Wnt signaling controls the activity of certain genes and regulates the interactions between cells during embryonic development.

The protein produced from the *WNT10A* gene plays a role in the development of many parts of the body. It appears to be essential for the formation of tissues that arise from an embryonic cell layer called the ectoderm. These tissues include the skin, hair, nails, teeth, and sweat glands. Researchers believe that the WNT10A protein is particularly important for the formation and shaping of both baby (primary) teeth and adult (permanent) teeth.

Health Conditions Related to Genetic Changes

Hypohidrotic ectodermal dysplasia

Several mutations in the *WNT10A* gene have been found to cause hypohidrotic ectodermal dysplasia, the most common form of ectodermal dysplasia. Starting before birth, ectodermal dysplasias result in the abnormal development of the skin, hair, nails, teeth, and sweat glands. Hypohidrotic ectodermal dysplasia is characterized by a reduced ability to sweat (hypohidrosis), sparse scalp and body hair (hypotrichosis), and several missing teeth (hypodontia) or teeth that are malformed. *WNT10A* gene mutations account for about 5 percent of all cases of hypohidrotic ectodermal dysplasia.

Most of the *WNT10A* gene mutations associated with hypohidrotic ectodermal dysplasia change single protein building blocks (amino acids) in the WNT10A protein, which impairs its function. The resulting shortage of functional WNT10A protein disrupts Wnt signaling during the development of ectodermal tissues, particularly the teeth.

Hypohidrotic ectodermal dysplasia can result from mutations in several genes. When the condition is caused by *WNT10A* gene mutations, its features are more variable than when the condition is caused by mutations in any other gene. Signs and symptoms range from mild to severe, and mutations in this gene are more likely to cause all of the permanent teeth to be missing.

Keratoconus

Other disorders

Mutations in the *WNT10A* gene have been reported to cause several other, rare forms of ectodermal dysplasia, including odonto-onycho-dermal dysplasia (OODD) and Schopf-Schulz-Passarge syndrome (SSPS). OODD is characterized by dry hair, missing teeth, a smooth tongue, fingernail and toenail abnormalities, thickened skin on the palms of the hands and soles of the feet (palmoplantar keratoderma), and increased sweating (hyperhidrosis) of the palms and soles. The major features of SSPS include missing teeth, hypotrichosis, palmoplantar keratoderma, and fluid-filled sacs (cysts) on the edges of the eyelids.

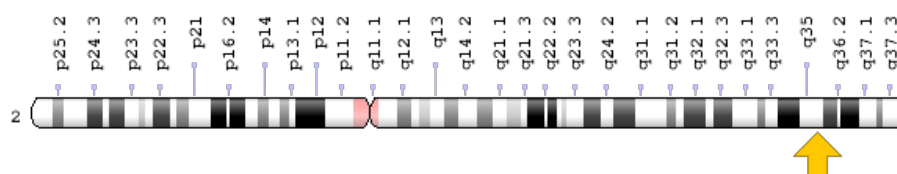
WNT10A gene mutations are also frequently associated with a condition called nonsyndromic tooth agenesis. This condition causes one or more teeth not to form. Although missing teeth is a common feature of ectodermal dysplasias, "nonsyndromic" suggests that in these cases tooth agenesis occurs without the other signs and symptoms of those conditions.

More than 70 *WNT10A* gene mutations have been identified in people with various forms of ectodermal dysplasia or nonsyndromic tooth agenesis. Most of these mutations change single amino acids in the *WNT10A* protein, which impairs its function. The resulting shortage of functional *WNT10A* protein disrupts Wnt signaling during the development of ectodermal tissues, including the skin, hair, nails, teeth, and sweat glands. Researchers are working to determine why mutations in this gene can cause several different disorders.

Chromosomal Location

Cytogenetic Location: 2q35, which is the long (q) arm of chromosome 2 at position 35

Molecular Location: base pairs 218,874,116 to 218,893,931 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- OODD
- protein Wnt-10a precursor

- STHAG4
- wingless-type MMTV integration site family, member 10A

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: The Wnt Gene Family and the Evolutionary Conservation of Wnt Expression
<https://www.ncbi.nlm.nih.gov/books/NBK6212/>
- The Wnt Homepage, Stanford University
<https://web.stanford.edu/group/nusselab/cgi-bin/wnt/>

Clinical Information from GeneReviews

- Hypohidrotic Ectodermal Dysplasia
<https://www.ncbi.nlm.nih.gov/books/NBK1112>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28WNT10A%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

Catalog of Genes and Diseases from OMIM

- ODONTOONYCHODERMAL DYSPLASIA
<http://omim.org/entry/257980>
- SCHOPF-SCHULZ-PASSARGE SYNDROME
<http://omim.org/entry/224750>
- TOOTH AGENESIS, SELECTIVE, 4
<http://omim.org/entry/150400>
- WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 10A
<http://omim.org/entry/606268>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_WNT10A.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=WNT10A%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:13829
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:80326>

- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/80326>
- UniProt
<https://www.uniprot.org/uniprot/Q9GZT5>

Sources for This Summary

- Adaimy L, Chouery E, Megarbane H, Mroueh S, Delague V, Nicolas E, Belguith H, de Mazancourt P, Megarbane A. Mutation in WNT10A is associated with an autosomal recessive ectodermal dysplasia: the odonto-onycho-dermal dysplasia. *Am J Hum Genet.* 2007 Oct;81(4):821-8. Epub 2007 Aug 9.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17847007>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1973944/>
- Arzoo PS, Klar J, Bergendal B, Norderyd J, Dahl N. WNT10A mutations account for ¼ of population-based isolated oligodontia and show phenotypic correlations. *Am J Med Genet A.* 2014 Feb;164A(2):353-9. doi: 10.1002/ajmg.a.36243. Epub 2013 Nov 25.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/24449199>
- Bohring A, Stamm T, Spaich C, Haase C, Spree K, Hehr U, Hoffmann M, Ledig S, Sel S, Wieacker P, Röpke A. WNT10A mutations are a frequent cause of a broad spectrum of ectodermal dysplasias with sex-biased manifestation pattern in heterozygotes. *Am J Hum Genet.* 2009 Jul;85(1):97-105. doi: 10.1016/j.ajhg.2009.06.001. Epub 2009 Jun 25.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19559398>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2706962/>
- Cluzeau C, Hadj-Rabia S, Jambou M, Mansour S, Guigue P, Masmoudi S, Bal E, Chassaing N, Vincent MC, Viot G, Clauss F, Manière MC, Touponay S, Le Merrer M, Lyonnet S, Cormier-Daire V, Amiel J, Faivre L, de Prost Y, Munnich A, Bonnefont JP, Bodemer C, Smahi A. Only four genes (EDA1, EDAR, EDARADD, and WNT10A) account for 90% of hypohidrotic/anhidrotic ectodermal dysplasia cases. *Hum Mutat.* 2011 Jan;32(1):70-2. doi: 10.1002/humu.21384.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20979233>
- Kantaputra P, Kaewgahya M, Jotikasthira D, Kantaputra W. Tricho-odonto-onycho-dermal dysplasia and WNT10A mutations. *Am J Med Genet A.* 2014 Apr;164A(4):1041-8. doi: 10.1002/ajmg.a.36388. Epub 2014 Jan 23.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/24458874>
- Mues G, Bonds J, Xiang L, Vieira AR, Seymen F, Klein O, D'Souza RN. The WNT10A gene in ectodermal dysplasias and selective tooth agenesis. *Am J Med Genet A.* 2014 Oct;164A(10):2455-60. doi: 10.1002/ajmg.a.36520. Epub 2014 Apr 3.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/24700731>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4167166/>

- Plaisancié J, Bailleul-Forestier I, Gaston V, Vaysse F, Lacombe D, Holder-Espinasse M, Abramowicz M, Coubes C, Plessis G, Faivre L, Demeer B, Vincent-Delorme C, Dollfus H, Sigaudy S, Guillén-Navarro E, Verloes A, Jonveaux P, Martin-Coignard D, Colin E, Bieth E, Calvas P, Chassaing N. Mutations in WNT10A are frequently involved in oligodontia associated with minor signs of ectodermal dysplasia. *Am J Med Genet A*. 2013 Apr;161A(4):671-8. doi: 10.1002/ajmg.a.35747. Epub 2013 Feb 7.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/23401279>
- Tardieu C, Jung S, Niederreither K, Prasad M, Hadj-Rabia S, Philip N, Mallet A, Consolino E, Sfeir E, Noueiri B, Chassaing N, Dollfus H, Manière MC, Bloch-Zupan A, Clauss F. Dental and extra-oral clinical features in 41 patients with WNT10A gene mutations: A multicentric genotype-phenotype study. *Clin Genet*. 2017 Nov;92(5):477-486. doi: 10.1111/cge.12972. Epub 2017 Mar 19.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/28105635>

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